NCBDDDannualreport2011
National Center on Birth Defects and Developmental Disabilities • Centers for Disease Control and Prevention

Sickle Cell Disease and Thalassemia

Preventing and Controlling Complications from Hemoglobinopathies

What is the problem?

- People with sickle cell disease (SCD) have a 20-30 year lower life expectancy than people without SCD.
- People with SCD, especially infants and children, are at risk for harmful infections. Pneumonia is a leading cause of death in infants and young children with SCD.
- An estimated 90% of people with SCD are unable to attain the resources they need for good management of their disease.
- Proven therapies are not being fully utilized. It is estimated that 30% or less of eligible patients are treated with hydroxyurea, a disease-modifying therapy for SCD.
- Because of the need for frequent blood transfusions, people with thalassemia are at increased risk for exposure to transfusion-related infections.
- Because there is no natural way for the body to eliminate iron, the iron in the transfused blood cells used to treat people with thalassemia can build up - iron overload - and become toxic to tissues and organs, particularly the liver and heart**.**

What do we know?

- Sickle cell disease (SCD) is one of the most common inherited blood disorders in the United States, with an estimated 100,000 Americans living with SCD.
- SCD occurs when a person inherits an abnormal gene from both parents. If both parents carry the sickle cell gene, there is a 25 percent chance that their baby will be born with the disease.
- It is estimated that 3 million people in the United States carry one sickle cell gene, that is, have sickle cell trait (SCT).
- SCD is more common in people of African, Southeast Asian, and Mediterranean descent.

- There are several types of thalassemia. An estimated 1,000 people have Cooley's anemia (the most severe form of thalassemia) in the United States, and an unknown number are carriers – people who have the genetic trait and can pass it on to their children.

- Thalassemia is most common among people of Mediterranean descent, such as Italians and Greeks, and is also found among people from the Arabian Peninsula, Iran, Africa, Southeast Asia, and Southern China.

Did you know?

- Fewer than 10% of Americans with SCD have access to treatment centers that specialize in management of this disorder.
- Children with SCD can, and should, participate in physical activity to help stay healthy. However, it's important that they don't overdo it, rest when tired, and drink plenty of water, as is true for all children – those with and without SCD.
- Hydroxyurea has been reported to reduce pain crises, strokes, disability, and death in people with SCD.
- Some people with SCT have been shown to be more likely than those without SCT to experience heat stroke and muscle breakdown when doing intense exercise, such as competitive sports or military training under unfavorable temperatures (very high or low) or conditions. Studies have shown that the chance of this problem can be reduced by avoiding dehydration and getting too hot during training.
- Most patients with Cooley's Anemia, the most severe form of thalassemia, receive red blood cell transfusions every two to three weeks, amounting to as much as 52 pints of blood a year.

What can we do?

- Increase public awareness of risks for those with SCT and their children, and help with health-related decisions.

- Increase general awareness about SCD for the public and health care providers.

- Increase patients' and providers' awareness and knowledge about and use of the preventative measures proven to reduce secondary complications due to SCD such as vaccinations, early and continuous screening, and disease modifying therapies such as hydroxyurea.

- Increase efforts to improve SCD and thalassemia-related healthcare, education, and systems for delivering care through policy development, community engagement and partnerships.

Accomplishments

- Co-hosted with the Sickle Cell Disease Association of America, “World Sickle Cell Awareness Day-Educate and Unite.” The event highlighted the need to increase awareness of the global impact of SCD and the importance of uniting global support for promoting and improving the health of persons with SCD through "increasing global action to reduce child mortality in SCD.”

- Launched “Living Well with Sickle Cell Disease-a Self-Care Tool Kit” to help people living with SCD manage their health and keep track of important information regarding medical care and treatment. Included in the toolkit are tracking sheets for vaccinations, immunizations, and hospitalizations and health provider information.

- Began a public health webinar series on hemoglobinopathies (SCD and thalassemia) to offer a learning collaborative platform for providers, consumers, educators, and scientists. Presenters are experts from a variety of disciplines, including the social sciences, clinical medicine, public health and community-based organizations.

- Convened the “International Public Health Learning Collaborative on Hemoglobinopathies” for countries, states, organizations, and others interested in learning more about public health tracking for SCD and thalassemia.

- In partnership with the Cooley’s Anemia Foundation, conducted five town hall meetings with patients with thalassemia to better understand the patient’s perspective on the challenges to fully adhering to prescribed treatments for thalassemia.

- Completed the first-ever environmental scan on educational materials for people with thalassemia which will help expand public access to an up-to-date and relevant collection of information resources on thalassemia.

- Provided input to a committee of thalassemia doctors and representatives from the Cooley’s Anemia Foundation, and the American Academy of Pediatrics on its proposal to expand the International Classification of Diseases (ICD) codes for thalassemia from one to six allowing for a more accurate diagnosis for thalassemia patients and enhance public health tracking activities. The proposal was accepted by the U.S. governmental agencies responsible (National Center on Health Statistics and the Centers for Medicare and Medicaid Services) for overseeing all changes and modifications and became effective October 1, 2011.

Looking to the future

- Increase the focus on preventing and controlling complications resulting from hemoglobinopathies.

- Publish reports that will track incidence and demographic characteristics, mortality rates, and health care utilization of people with SCD.

- Fund 1-2 state health departments to establish a population-based tracking system for pediatric and non-pediatric populations with SCD to identify those receiving disease-modifying therapies, such as hydroxyurea, and SCD related secondary complications.

- Support the US Department Health and Human Services initiative to increase access and improve care of people with SCD by collecting information and conducting research to develop and disseminate evidence-based guidelines that will prevent and decrease complications associated with SCD.

- Partner with the American Society of Hematology to develop and launch a Sickle Cell Trait Education, Information and Resource Tool Kit that will provide general information about SCT (What is SCT, who's at risk) and information and resources concerning family planning, screening, genetic counseling, potential complications, prevention and management of complications, and psychosocial considerations. Information will be tailored to meet the education, information and resource needs of parents, children/adolescents/adults, health care providers, school coaches, teachers and administrators, athletes, state health departments, and US community advocates and health educators.

- Work with the Cooley's Anemia Foundation to develop a patient guide to living with thalassemia, a thalassemia toolkit for use in schools, and an emergency preparedness checklist specially designed for the thalassemia population.

Stories

Phyllis Zachery-Thomas

Phyllis Zachery-Thomas is 47 years old and was diagnosed with sickle cell disease at the age of six months old. She is a wife, mother, grandmother, and champion for the sickle cell community. She started the SCD Soldier Network, a non-profit organization [scdsoldiernetwork.com] whose mission is to support and mobilize the sickle cell community. Phyllis shared the following story with us:

"Sickle cell disease is unpredictable and you don't know when it will attack. Although people with sickle cell disease are living longer, it is a battle to continue living. The disease has waged war against us. If you aren't going to a specialty care system, you will likely find medical personnel who aren't trained to treat sickle cell disease. I've had to educate my doctors. It took a long time for me to manage it. I'm someone who has gone through every phase of sickle cell and I've seen its many faces."

Stories like Phyllis' underscore the importance to improve access, quality of care, and length and quality of life for people living with SCD.

Terese Finitzo

"My name is Terese Finitzo. This is a picture of me and my two sisters, with me on the far right. I'm ten years old in this picture; long after I had experienced the shattered glass-like pain in my arms and legs that I eventually learned came from sickle cell disease. (People who have this form of SCD inherit a sickle cell gene ("S") from one parent and from the other parent a gene for an abnormal hemoglobin called "C." This is usually a milder form of SCD.)

In the early years, the pain came mostly in the summer after swimming, though our family doctors told my parents there was nothing wrong with me; that I was spoiled. I was diagnosed with sickle cell disease when I was 12, but we were not told of the diagnosis. Indeed, many years later at age 24, I asked, what was the explanation for why I had become so ill on my first ski vacation? The original pediatric hematologist was still in practice and he said, 'Oh yes, you're the young redheaded child with Sickle C disease." The family doctors decided not to tell my Italian immigrant father and my Scotch Irish mother living on the South side of Chicago in the late 1950's that their daughter had a disease found "only" in those of African ancestry.

We eventually learned my Mediterranean father carried the C trait and my mother, whose family had been in the US for nearly 200 years carried the S trait. They met during the war in California, and moved back to Chicago where they had three daughters. I am the only one with the disease, though my two sisters lived with the pain of seeing me suffer then and still do today when they see me fall ill.

It is time that there is universal newborn screening for sickle and other hemoglobinopathies, and also for identifying individuals with trait."

Terese's story is a poignant reminder of the many faces of SCD and that there is still a lot of work to be done to educate the public and health care providers about SCD.

Notable 2011 NCBDDD Scientific Publications

- Grant AM, Parker CS, Jordan L, Hulihan M, Creary M, Lloyd-Puryear MA, Goldsmith JC, Atrash H. Public health implications of sickle cell trait: A Report of the Centers for Disease Control and Prevention Meeting. American Journal of Preventive Medicine 2011;41(6S4):S435-39.

- Grosse SD, Odame I, Atrash HK, Amendah D, Piel FB, Williams TN. Sickle cell disease in Africa: a neglected cause of early child mortality. American Journal of Preventive Medicine 2011;41(6S4):S398-S405.
- Hinton CF, Grant AM, Grosse SD. The ethical and practical implications of ethnically-targeted screening for genetic disorders: The case of hemoglobinopathy screening. Ethnicity & Health. 2011;16(4-5): 377–388.
- Jordan LB, Smith-Whitley K, Treadwell MJ, Telfair J, Grant AM, Ohene-Frempong K. Screening U.S. College Athletes for Their Sickle Cell Disease Carrier Status. American Journal of Preventive Medicine 2011;41(6S4):S406-12.
- Swanson ME, Grosse SD, Kulkarni R. Disability and sickle cell disease: A framework for public health assessment. American Journal of Preventive Medicine 2011;41(6S4):S390-97.
- Yusuf H, Lloyd-Puryear MA, Grant AM, Parker CS, Creary MC, Atrash HK. Sickle cell disease: the need for a public health agenda. American Journal of Preventive Medicine 2011;41(6S4):S376-83.